Claims

1 A method for the diagnosis of a polymorphism in a PDH E1β gene in a human, which method comprises determining the sequence of the nucleic acid of the human at one or more of positions 457, 1191, 1198 and 1342 in the PDH E1β gene as defined by the positions in SEQ ID NO: 1; and determining the status of the human by reference to polymorphism in the PDH E1β gene.

2 A method according to claim 1 in which the polymorphisms are further defined as:

Position	Polymorphism	Region
457	A/G	Coding, silent Gly
1191	A/C	3'UTR
1198	C/T	3'UTR
1342	C/A	3'UTR

3 A method according to claim 2 which comprises diagnosis of any one of the following haplotypes:

(a) 1191C 1198C 1342A;

(b) 1191A 1198C 1342C; or

(c) 1191C 1198T 1342A.

4 An isolated nucleic acid comprising the nucleic acid of SEQ ID NO: 1 with C at position 1191 as defined by the position in SEQ ID NO: 1; or a complementary strand thereof or an antisense sequence thereto or a fragment thereof of at least 20 bases comprising C at position 1191.

5 An allele specific primer capable of detecting a PDH E1β gene polymorphism at one or more of positions 457, 1191, 1198 and 1342 in the PDH E1β gene as defined by the positions in SEQ ID NO: 1.

6 An allele-specific oligonucleotide probe capable of detecting a PDH E1β gene polymorphism at one or more of positions 457, 1191, 1198 and 1342 in the PDH E1β gene as defined by the positions in SEQ ID NO: 1.

7 Use of any polymorphism as defined in claim 2 as a genetic marker in a linkage study.

8 A method of treating a human in need of treatment with a PDH drug in which the method comprises:

i) diagnosis of a polymorphism in the PDH E1β gene in the human, which diagnosis comprises determining the sequence of the nucleic acid at one or more of positions 457, 1191, 1198 and 1342 in the PDH E1β gene as defined by the positions in SEQ ID NO: 1, and determining the status of the human by reference to polymorphism in the PDH E1β gene; and

ii) administering an effective amount of a PDH drug.

9 Use of any one of the following in bioinformatic analysis:

i) any polymorphism defined in claim 1 or 2; or

ii) any haplotype defined in claim 3.

10 A use according to claim 9 comprising a bioinformatic analysis selected from homology searching, mapping, haplotyping, genotyping or pharmacogenetic.